Cystic Fibrosis

In November 2007, the Arizona Office of Newborn Screening began screening for Cystic Fibrosis (CF). The addition of CF screening completed the nationally recommended panel of 29 disorders, including hearing loss. This is a great achievement for Arizona.

Description of Cystic Fibrosis

CF is an inherited disorder that is treatable and not contagious. CF causes thick, sticky mucus to build up in the lungs and other organs. A baby with CF will need regular medical care and a good diet. One in every 3,500 babies is born with cystic fibrosis.

Babies with CF often do not have visible signs of disease for weeks, months, or years. New treatment has improved the quality and length of life for CF patients. Babies should not be limited in their future goals and plans.

Symptoms of CF may include: persistent coughing, wheezing or shortness of breath, recurrent respiratory infections, upset stomach, and excessive appetite but poor weight gain.

Process of Cystic Fibrosis Screening

Arizona aims to receive two blood specimens from each newborn in the state. Each first specimen for a newborn is screened for Cystic Fibrosis using the following process (a small percentage of specimens cannot be tested):

1. Blood specimen is collected by a trained professional
2. Specimen is sent to Arizona's contracted laboratory for testing
3. Specimen is screened for Immunoreactive Trypsinogen (IRT)
    a. Cutoff is the top 2.2% of values for the day
    b. Values below the top 2.2% are considered normal and not indicative of CF
4. Specimens with IRT values in the top 2.2% are screened for a panel of the 46 most common DNA mutations associated with CF
    a. Possible outcomes: No mutations, one mutation, or two mutations found
    b. No mutations means CF is not indicated
    c. One mutation means CF may be present but diagnostic testing is needed
    d. Two mutations means CF is likely but diagnostic testing is needed
5. Patients with results indicating possible CF are referred for an evaluation and diagnostic testing at Arizona's two Cystic Fibrosis Centers
    a. The current "gold standard" diagnostic test for CF is commonly called a sweat test
    b. Arizona's two Cystic Fibrosis Centers are:
        i. Phoenix Children's Hospital
        ii. Arizona Respiratory Center in Tucson (University Medical Center)
6. Office of Newborn Screening works closely with the Cystic Fibrosis Centers to encourage families of newborns who have abnormal results to receive follow-up appointments with qualified specialists. The Office administers the education and quality aspects of CF screening.

For more information, please see our website at www.AZNewborn.com

46 DNA Mutations Which Can Be Detected by Arizona Cystic Fibrosis Screening

ACOG/ACMG:
ACMG-23
1078delT
l148T

Reflex:
5T
7T
9T
F508C

Additional Select Analytes:
2183A A>G
3849+4 A>G
3876delA *
3905insT
394delTT
D1152H
D1270N
E60X
Q493X
R347H
S549N *
S549R A>G
S549R T>G
V520F
Y1092X C>A
Y1092X C>G
Y122X

ACMG-23
delF508
G542X
G551D
W1282X
3849+10kb C>T
N1303K
621+1 G>T
1717-1 G>A
R553X
R117H
G85E
2789+5 G>A
3120+1 G>A
delI507
R1162X
1898+1 G>A
3659delC
711+1 G>T
R334W
2184delA
A455E
R347P
R560T